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Neurological and dysmorphic features in a family with brachial plexus neuropathy. *A. Chen, M. Kovach, V. Kimonis.* Div. of Genetics & Metabolism, Dept Pediatrics, Southern Illinois Univ, Springfield, IL.

Hereditary neuralgic amyotrophy (MIM 162100) or familial plexus neuropathy is an autosomal dominant disorder characterized by the onset of recurring episodes of back pain and muscle weakness early in childhood. Affected individuals also have hypotelorism or closely spaced eyes. Although the molecular basis for this disorder remains unknown, genetic linkage has been established to chromosome 17q24-q25 (Pellegrino et al. 1997; Stogbauer et al. 1997). We report a four-generation family presenting with autosomal dominant brachial plexus neuropathy. The proband a 16 y. old male presented with pain and atrophy of his left deltoid, suprascapular, rhomboid muscles (causing winging of his scapular) and abductor pollicis brevis muscle. A further episode of pain in his right arm did not lead to permanent loss of function. Dysmorphic evaluation in him revealed hypotelorism and small palpebral fissures. His father age 47 y. was asymptomatic, however had hypotelorism. His paternal uncle age 49 y. has had a history of recurrent episodes of pain preceding weakness in his arms since the age of 17 y. He has marked wasting of the muscles in his hands and similar facies. The paternal grandmother developed recurrent paralysis in both arms since a pregnancy at the age of 24 y. She also developed weakness of her legs, became bedbound this leading to her death at the age of 54 y. Genetic linkage analysis with chromosome 17q24-q25 markers was performed in six family members (2 affected, 1 carrier, 2 unaffected, and 1 spouse). The power of the pedigree was not sufficient to generate significant LOD scores (maximum LOD score 0.109 at a recombination fraction = 0.00 for marker D17S802), however haplotype analysis with markers D17S802, D17S939 and D17S1603 revealed a shared haplotype among affected individuals. This study further confirms the lack of heterogeneity of this interesting familial disorder causing facial dysmorphisms and recurrent episodes of neuropathy.

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